Rare Kids

Individually rare, collectively common

A guide for families navigating a rare condition

The Rare Condition Passport and Toolkit



Rare Diseases NSW

Navigating life with a rare condition

Dear parents, families and carers,

Caring for a child with a rare condition can be challenging. It can also feel confusing and lonely at times. Families find themselves learning new medical terms, meeting different health professionals, and trying to make sense of what comes next.

Whether your child has recently been diagnosed, you are still looking for answers, or you have been on this path for some time, this resource is here to support you.

Inside, you will find simple information about rare conditions, the healthcare system, support services, and family resources. You will also find ways to connect with other families and organisations who understand these experiences.

Our understanding of rare diseases has come a long way. Through research and listening to families' experiences, we are continuing to learn more about rare diseases and how to better support children and families.

We hope this guide helps you feel less alone and more supported in what comes next.

Warm regards,

Nika Kulaweera,
Rare Kids Nurse Specialist
The Sydney Children's Hospitals Network

Jessie Ballentine,
Rare Kids Social Worker
The Sydney Children's Hospitals Network

Rare Diseases NSW and the Rare Kids and Gene2Care teams at Sydney Children's Hospitals Network



Acknowledgements

Rare Diseases NSW respectfully acknowledges the Traditional Custodians of the lands throughout Australia.

We acknowledge and honour the enduring connection of Aboriginal and Torres Strait Islander peoples to this land, to culture, to community and to Country. We pay our respects to the people, the cultures and the Elders past and present.

Acknowledgement of Lived Experience

Rare Diseases NSW acknowledges the people living with rare diseases, their families, carers and communities. Your lived experience shapes everything we do, including this guidebook.

The expertise you have gained through your lived experiences is invaluable. Your strength, ideas, and leadership help improve care, research, education, and policies.

We are committed to working together and listening to your experiences. We value your knowledge just as much as medical and research knowledge. Together, we are creating a fairer, more person-centred future, where everyone living with a rare disease is heard, respected, and supported.

These materials have been developed as part of The Rare Condition Passport and Toolkit funded by the Australian Government.

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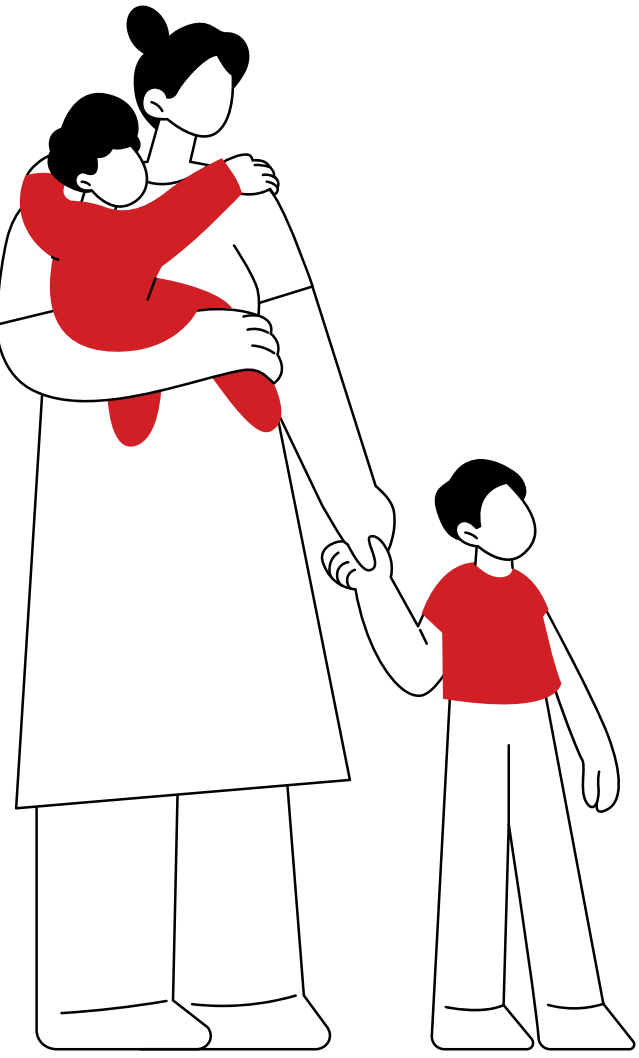
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What is a rare condition?

A rare condition is one that affects fewer than 1 in 2,000 people. There are more than 9,000 known rare conditions. About 8 in 10 are caused by changes in a person's genes. Others can be caused by rare cancers, infections, or differences in the body's immune system. Examples include cystic fibrosis, Kleefstra syndrome, PURA-related condition, fragile X syndrome, Zika virus, SCN2A-related conditions, and juvenile dermatomyositis syndrome. While each condition is rare on its own, together they are common. Around 2 million Australians live with a rare condition. [1,2,3,4]

*Please see **Genetics 101** at the end of this document for a brief explanation of DNA, genes, and chromosomes, and a list of reliable resources for more information about genetics and rare conditions.*





Your healthcare team

Every child with a rare condition is unique. The healthcare professionals on your child's team will be different based on your child's needs, their condition and where you live. Australia has national recommendations for rare disease care. They say your child should get coordinated care for life, with your GP at the centre of the team of healthcare professionals. [4]

It is also important to know you can ask for a **second opinion** if you did not feel your questions are being taken seriously. You have the right to clear information, to take time to make decisions (unless it is an emergency), and to ask for copies of your child's medical records. Every person in Australia has health care rights.

These are explained in the Patient Charter:

safetyandquality.gov.au/your-rights/understanding-your-healthcare-rights

Your core team

You and your child

It is important to note that healthcare professionals will not have a full understanding of each rare condition. Often families will need to educate healthcare professionals about their child's condition.

As a family, you are the expert when it comes to your child's care.

Paediatrician

A children's doctor who:

- oversees general health, growth and development
- helps plan and coordinate your child's care.

GP (family doctor) and/or and or Aboriginal or Torres Strait Islander Controlled Health Centre doctor

Your doctor:

- looks after general health and wellbeing
- makes referrals to specialists
- provides prescriptions
- requests investigations
- provides care coordination and support for families.

Your wider team

Based on your child's needs, your team may also include:

Allied health

- Physiotherapy (help with movement and strength)
- Occupational therapy (help with everyday skills)
- Speech pathology (help with talking and communication)
- Dietetics (help with food and growth).

Mental health

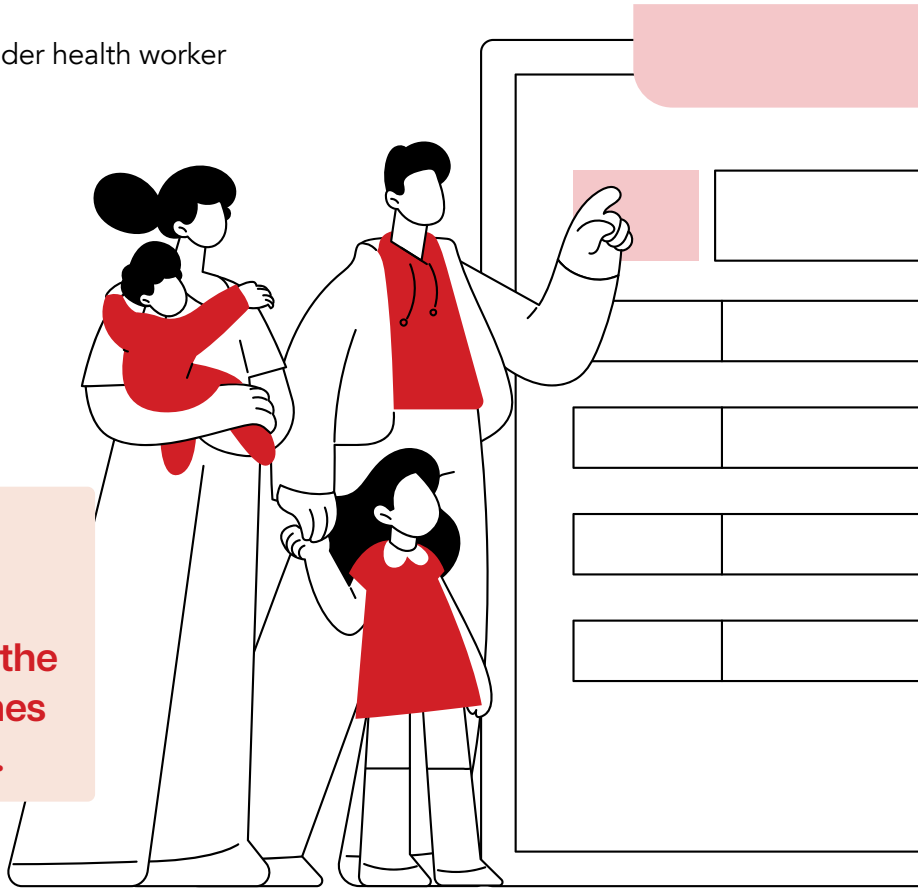
- Psychologist
- Psychiatrist
- Neuropsychologist (helps with thinking, learning and behaviour).

Sub-specialty teams

- Geneticist (diagnoses rare genetic conditions gives advice on new treatments)
- Neurologist (brain and nerves)
- Gastroenterologist (stomach and gut)
- ENT, also called Otolaryngologist (ears, nose and throat)
- Sleep physician.

Community support

- Family and friends
- Support groups
- School
- Disability support person
- Aboriginal and/or Torres Strait Islander health worker
- Multicultural health worker.



As a family, **you are the expert when it comes to your child's care.**

Specialist health care supports for people from priority populations

Everyone deserves health care that fits who they are. Some families can get extra help to make care easier to reach and easier to understand. Here are some supports you can ask about, wherever you live in Australia.

First Nations families

If you are Aboriginal or Torres Strait Islander, a free program called Integrated Team Care can give you a Care Coordinator who works with you, your doctor and other services to help you manage your child's care. They can help you understand the condition, book appointments, and in some areas help with transport. Aboriginal Community Controlled Health Services offer care that is run by and for community. Ask your doctor to refer you, or find a service near you through NACCHO: naccho.org.au

Families of children with disability

Some hospitals have a disability liaison officer who can make visits easier, and some areas have care navigators who help you find and join up services. It is fine to ask your team what is available near you. The NDIS can also connect you with a Local Area Coordinator, or an Early Childhood Partner for younger children, who help you find services and apply if needed (see Chapter 6). Call the NDIS on 1800 800 110 or visit: ndis.gov.au

Children with intellectual disability

People with intellectual disability can ask their GP for a yearly health check. This is a longer appointment that looks at overall health and helps find problems early. In some states there are also specialist intellectual disability health teams that work with your GP to make health care plans and find the right services. Ask your GP what is available where you live. You can also find mental health and wellbeing information at: idmhconnect.health

Families who speak a language other than English

You have the right to a free, private interpreter when you use health services. You can ask for one at any time, and you never have to pay. Even if you go with a family member or friend who speaks English, a trained health interpreter is advised as they can explain complex medical words and concepts in a way everyone can understand.

An interpreter helps you and your doctor understand each other clearly. Tell the clinic or hospital before your visit, or call the Translating and Interpreting Service on 131 450.

tisnational.gov.au

Families in regional, rural and remote areas

If you live a long way from a specialist, you may need to travel for care. Every state and territory has a travel assistance scheme that helps with the cost of travel and accommodation when the care you need is not available close to home. You apply in the state or territory where you live. Ask your hospital social worker or your doctor to help you apply. Telehealth (video appointments) can also save some trips.

- **New South Wales:** IPTAAS, iptaas.health.nsw.gov.au
- **Victoria:** VPTAS, health.vic.gov.au
- **Queensland:** PTSS, health.qld.gov.au/ptss
- **South Australia:** PATS, sahealth.sa.gov.au/pats
- **Western Australia:** PATS (WA Country Health Service)
- **Tasmania:** PTAS
- **Northern Territory:** PATS
- **Australian Capital Territory:** IPTAS

For families in Western Australia, Tasmania, the Northern Territory and the Australian Capital Territory, you can find the scheme on your state or territory health department website, or your hospital social worker can help. Families in remote areas can also reach the Royal Flying Doctor Service.

flyingdoctor.org.au

Building a support network

Linking with other families with similar experiences can help. The following supports and resources may be helpful as you navigate life with a rare condition. They share information and can link you with peer support.

Genetic Alliance Australia.

Provides information, peer support and connections for families affected by rare genetic and undiagnosed conditions. They can link people to support groups and services, even when no group exists for a specific condition.

geneticalliance.org.au

Belongside Families.

A family-led not-for-profit offering free programs, peer support groups, workshops and resources for families raising children with disability, developmental delay, and rare or genetic conditions.

belongsidefamilies.org.au

MyTime.

Free, facilitated peer support groups where parents and carers of children with disability, developmental delay or chronic conditions can connect, share experiences and build confidence.

mytime.net.au

Rare Voices Australia.

The national peak body for Australians living with a rare condition. The RARE Portal gives families step-by-step guidance on what to do if a rare disease is diagnosed or suspected.

rarevoices.org.au

The Rare Disease Disability Toolkit.

- Community (inclusion and confidence in daily life),
- Health (self-advocacy tools for the disability/health interface), and
- The National Disability Insurance Scheme (navigating the NDIS for better outcomes).

rarevoices.org.au/toolkit-page/

First Peoples Disability Network.

First Peoples Disability Network (FPDN) is a national organisation run by and for Aboriginal and Torres Strait Islander people with disability and their families. They offer advocacy, information and connection.

fpdn.org.au/

Multicultural Disability Support Guide.

Families from many cultures and languages can find services that understand their needs. The Disability Support Guide lists multicultural disability services across Australia.
Remember you can always ask for a free interpreter at health appointments (call 131 450).

disabilitysupportguide.com.au/information/article/multicultural-services

Other groups that may help:

- Individual support groups may be available for your child's condition. You can find these online or through Genetic Alliance Australia or Rare Voices Australia.
- Informal support groups on Facebook or other social media. Because many conditions are rare, some support groups can only be found in Facebook groups. These groups are not usually run by health professionals, so use your own judgement when reading medical information shared in them. They can still offer helpful social connection.



Mental health supports

Mental health support for parents and carers

Caring for a child with a rare condition can affect your mental health and your physical wellbeing. Long-term stress may lead to carer burnout, anxiety, low mood, sleep problems or feeling overwhelmed. [4]

Looking after yourself is not selfish. It helps protect your mental health and your ability to care for your child.

The RARE Portal has resources about mental health supports and services for Australians living with rare conditions, their carers and their families.

rareportal.org.au

Carer Gateway (1800 422 737) provides free counselling, coaching, peer support and respite, including emergency respite.

carergateway.gov.au/

WellMob brings together social, emotional and cultural wellbeing online resources for Aboriginal and Torres Strait Islander people.

wellmob.org.au/

Intellectual Disability Mental Health Connect helps people with intellectual disability, and their family or support people find information on mental health and wellbeing.

idmhconnect.health/

Embrace Multicultural Mental Health provides multicultural communities with access to resources, links to services and translated information on mental health.

embracementalhealth.org.au/

Helpful strategies:

- take regular short breaks
- accept practical help
- stay connected with people who support you
- prioritise sleep and gentle movement
- speak with your GP, a counsellor, or ask about respite support
- ask your GP for a Mental Health Care Plan (10 sessions subsidised by Medicare).

A Mental Health Care Plan gives you up to 10 sessions a year with a psychologist, social worker or other mental health professional, partially or fully paid for by Medicare.

How to access:

- book a longer appointment with your GP
- ask for a mental health care plan
- talk openly about how caring is affecting you
- your GP can write the plan and refer you.

Carer Gateway is an Australian Government program providing free services and support for carers.

If you are looking after a child with a rare condition you are a carer and eligible for their free local support including phone advice, counselling and support to get respite.

carergateway.gov.au/

Support is available. You do not have to manage alone.

Looking after yourself is not selfish. It helps protect your mental health and your ability to care for your child.



Mental health support for siblings

Brothers and sisters are on this journey too, often quietly. They may feel confused, worried, guilty or upset, and they often hide it to protect you.

Parents and carers can help in simple ways:

- **Be honest in simple words.** When children do not understand, they fill in the gaps with their imagination. This can be scarier than the truth. You do not need to share every detail. Just enough to help them understand what is happening, and that it is not their fault. Ask what they want to know.
- **Protect some regular one-on-one time.** It does not need to be much. A weekly slot with your phone away does more than the odd big day out.
- **Help them explain to friends.** Not knowing what to say to friends can feel isolating. Help them find a simple, comfortable way to describe things.
- **Do not rush to fix their bad days.** Sometimes they just need to feel their feelings and be heard.
- **Mental health support.** Brothers and sisters may also benefit from speaking with a mental health professional. A GP can give them a Mental Health Care Plan for up to 10 sessions subsidised by Medicare.

For more support:

Siblings Australia offers resources, support programs, and information for siblings of people with disability or chronic illness, their families, and the professionals who support them.

siblingsaustralia.org.au

Young Carers Network has resources about support services and information for young Australians who care for a family member or friend with disability, mental illness, or chronic condition.

youngcarersnetwork.com.au



Accessing the NDIS

The National Disability Insurance Scheme (NDIS) provides funding and support for children with permanent and significant disability. You do not need a formal diagnosis to make an enquiry. *[4]*

Early childhood (under 9 years)

Children younger than 6 with a developmental delay, and children younger than 9 with a disability, can access support through an Early Childhood Partner.

Early Childhood Partners can:

- provide short-term early supports
- connect you with local services
- help you apply for the NDIS if needed
- support you through the planning process.

To get started:

- call the NDIS on 1800 800 110
- ask to be connected to your local Early Childhood Partner if your child is under 9.

Children 9 years and over: Local Area Coordinator Support

Children aged 9 and over can access the NDIS through a Local Area Coordinator.

They can:

- help you understand eligibility
- support you to gather reports and evidence
- help with NDIS planning
- connect you to community supports.

Contact the NDIS on 1800 800 110 or visit **ndis.gov.au** to find your local partner.

If your NDIS plan is not meeting your needs

You can ask for a plan review if the supports no longer meet your child's needs, or if your child needs new therapies, equipment or services. Contact the National Disability Insurance Agency (NDIA), your Local Area Coordinator, or your support coordinator to start the review.

The NDIS is the program that gives funding and support to people with disability so they can get the help they need in their daily lives.

The NDIA is the government organisation that runs the NDIS. It plans, manages and makes decisions about how the program works.

The Rare Voices Australia Disability Toolkit offers practical information on disability and rare disease and navigating the NDIS.

The Toolkit was codesigned with people living with rare conditions and facilitated by Rare Voices Australia. It brings together resources on:

- Community (inclusion and confidence in daily life),
- Health (self-advocacy tools for the disability/health interface), and
- The National Disability Insurance Scheme (navigating the NDIS for better outcomes).

Each resource includes a main guide plus supporting tools to help people speak up for themselves and work with their support team.

The RARE portal also has community information on disability supports for families from First Nations and culturally and linguistically diverse communities.

rarevoices.org.au/toolkit-page

Other money supports you can use with or instead of the NDIS

Toy libraries, Independent Living Centres and local disability services often loan equipment so you can try before you buy.

'Specialist health care supports for people from priority populations' lists the organisations which can offer tailored financial advice to First Nations families, families of children with disability, families who speak a language other than English, and families living in regional, rural, and remote areas.

Variety, the Children's Charity helps with equipment and therapy costs.

variety.org.au

John Maclean Foundation provides wheelchairs and mobility equipment.

johnmacleanfoundation.org

Steve Waugh Foundation supports families living with rare diseases.

stevewaughfoundation.com.au

Cure Kids helps with medical costs that are not covered elsewhere.

curekids.org.au

General support and information for parents and carers

The RARE Portal

Resources about mental health supports and services for Australians living with rare or undiagnosed conditions, their carers and their families.

rareportal.org.au

Carer Gateway

Free support for carers, including:

- counselling
- peer support
- coaching
- respite options
- emergency respite

Phone: **1800 422 737**

carergateway.gov.au

Siblings Australia

Support for brothers and sisters of children with disability or chronic illness. Offers:

- information and resources
- workshops
- support programs
- parent guidance on supporting siblings

siblingsaustralia.org.au

Young Carers Network

For children and young people who help care for a family member. Provides:

- information and advice
- school support guidance
- an online community
- links to local supports

youngcarersnetwork.com.au

Belongside Families

A family-led not-for-profit, by families for families raising children with disability, developmental delay, autism, and rare or genetic conditions. Offers:

- peer support groups
- education and information sessions
- webinars and workshops
- in-person catch ups
- 1:1 support tailored to your needs

belongsidefamilies.org.au

Kiind

Supports families raising children living with disability, autism and developmental delay. They offer practical assistance, emotional support and connection to other families to help find the most appropriate services and supports for children and their families.

Although Kiind is based in Western Australia, anyone across Australia can access their online resources.

kiind.com.au/

Advocating for children with disability (ACD)

A family-led organisation for families of children with disabilities. Provides information to help families raise a child with disability and developmental delay, from early years through to teenage years.

Although ACD is based in Victoria, anyone across Australia can access their online resources.

acd.org.au/

First Peoples Disability Network

First Peoples Disability Network (FPDN) is a national organisation run by and for Aboriginal and Torres Strait Islander people with disability and their families. They offer advocacy, information and connection.

fpdn.org.au/

Carer Gateway

Carer Gateway also has a First Nations fact sheet with support for carers.

carergateway.gov.au/resource/first-nations-fact-sheet

Day-to-day financial supports

Centrelink payments

Carer Allowance

Fortnightly payment for parents who give daily care to a child with disability or medical needs. Income tested.

servicesaustralia.gov.au/carer-allowance

Carer Payment

Income support for carers who cannot work full-time because they are caring for someone. Income tested.

servicesaustralia.gov.au/carer-payment

Carer Adjustment Payment

A one-off payment for families in money trouble after a sudden diagnosis or major event (for children under 7).

servicesaustralia.gov.au/carer-adjustment-payment

Charities

Steve Waugh Foundation

Provides grants for specialised equipment, therapy and other essential needs for children and adults with rare conditions.

stevewaughfoundation.com.au

Brainwave

Practical and financial support for children with brain conditions.

brainwave.org.au

Variety Australia

Therapy grants of up to $2,000. Helpful for families who may not be eligible for the NDIS.

variety.org.au

Research and future directions

For rare conditions, there is often less information and data than for common conditions. This makes research very important for the care of people living with a rare disease, both now and in the future.

Rare disease research can take different forms:

- Clinical trials, which test new treatments. Some look at advanced therapies, which are medicines made from genes, tissues or cells. For many people, a clinical trial is the only way to reach a treatment.
- Undiagnosed disease programs, which help find a diagnosis.
- Studies that are not about treatment, such as research into quality of life or how services are used.

Families can talk to their doctor about research that may suit their child and family. The RARE portal (rareportal.org.au/) also has information about clinical research. A rare condition support group is often a great place to find out about research too.

Here are some resources that can help.

Finding clinical trials

Australian Clinical Trials

This Australian government website lets people search for clinical trials being run in Australia and New Zealand. People can also set an alert, so they hear when new trials start.

australianclinicaltrials.gov.au/participants

Consumer Guide to Clinical Trials

Explains what a clinical trial is, some of the possible benefits and risks, and questions to ask when thinking about joining a trial.

involvementtoolkit.clinicaltrialsalliance.org.au/media/z2odqoly/cip-final.pdf

Kids Research

This website from the Sydney Children's Hospitals Network shows how to search for trials on the Australian New Zealand Clinical Trials Registry.

kr.schn.health.nsw.gov.au/clinical-trials/enrolling

ClinicalTrials.gov

A large database of clinical trials in the USA and around the world.

clinicaltrials.gov/

Other resources

Advanced Therapies Handbook

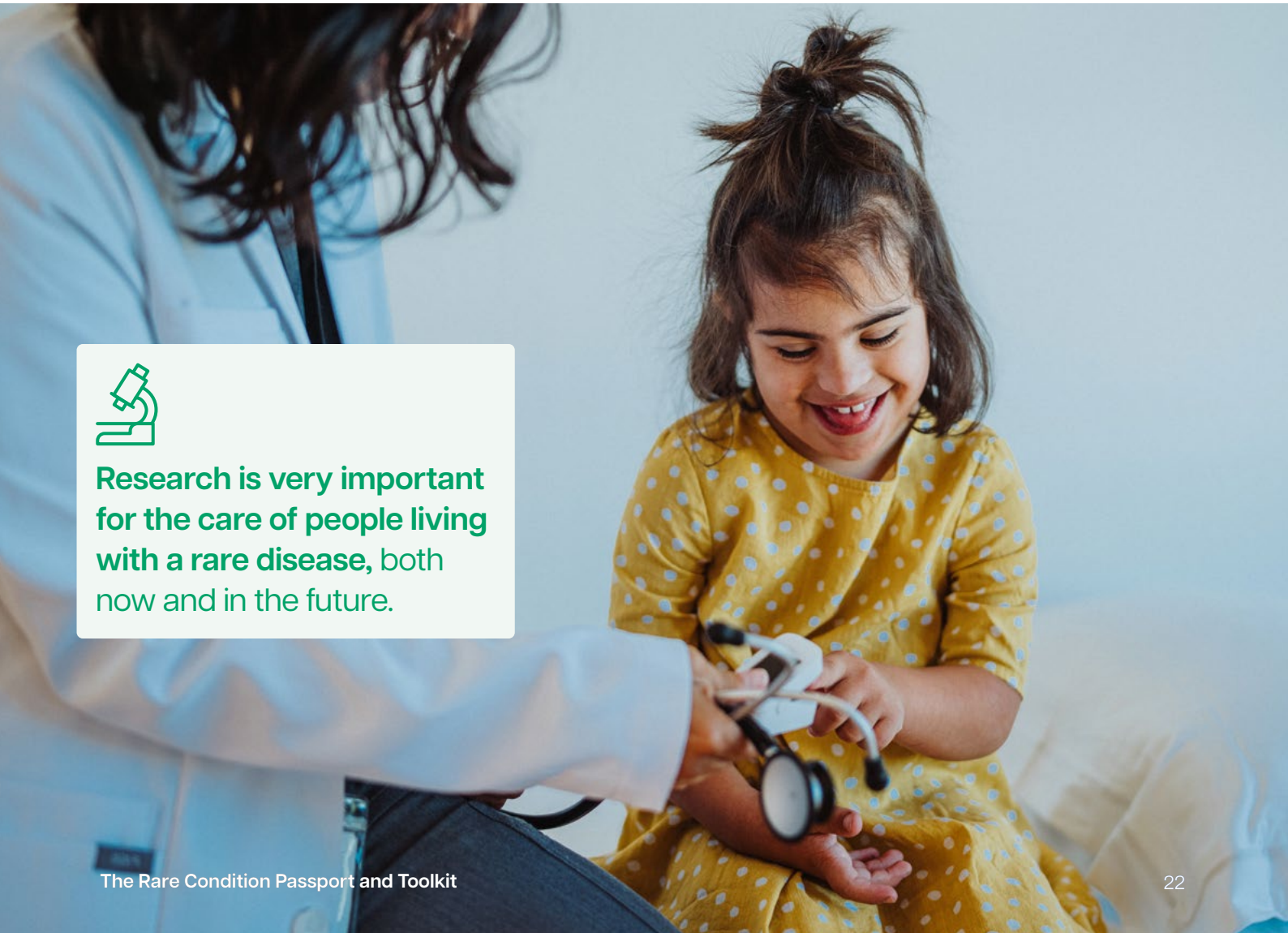
An Australian resource from the Sydney Children's Hospitals Network. It helps families and health professionals understand new and emerging advanced therapies used to treat rare diseases, including the journey from diagnosis to treatment.

schn.health.nsw.gov.au/advanced-therapies-handbook

Genomics of Rare Disease Registry

Part of a national study that looks at the genetic causes of rare diseases, so they can be better understood, found and managed. The registry aims to connect families affected by an inherited rare disease with new research opportunities, including studies that can help find a genetic diagnosis.

garvan.org.au/research/clinical-trials/rare-disease-registry



Research is very important for the care of people living with a rare disease, both now and in the future.

Research in New South Wales

Rare Diseases NSW

An Australian centre of expertise that brings together care, research, education and advocacy. It works to improve diagnosis, support and treatment for children, adults and families with rare and undiagnosed conditions. Families can take part in research, attend events, join the Community of Practice (a group of healthcare professionals and families working towards a common goal in rare disease care), share your story or support advocacy.

rarediseasesnsw.au

Gene2Care

Gene2Care brings research and clinical care together to improve care for children and families living with a diagnosed or suspected rare genetic condition. It has two parts: the GeneSTART registry and the GeneAdd undiagnosed disease program. Gene2Care is part of Rare Diseases NSW.

GeneAdd is a research program for people who may have a rare genetic condition but do not yet have a diagnosis. It helps families take part in research that may lead to a diagnosis faster. It also supports research into new treatments. GeneAdd is currently at the Sydney Children's Hospitals Network, Westmead Hospital and St George Hospital, and is expanding to Liverpool Hospital.

GeneStart is a rare disease registry. It supports families living with a rare condition by giving up-to-date information, links to resources, chances to connect with other families, and access to research. GeneStart also collects feedback from families and clinicians about their experiences with clinical genetics services. This helps improve care and reduce some of the uncertainty that can come with rare disease journeys.

kr.schn.health.nsw.gov.au/our-research/projects-and-initiatives/gene2care



Research in Victoria

Rare Disease Now (RDNow)

RDNow uses advanced tools to find a diagnosis for children who are still undiagnosed. This gives these children the best chance of getting a diagnosis and reaching the latest clinical trials and treatments.

mcri.edu.au/research/projects/rdnow

Research in Western Australia

The Rare Care Centre

The Rare Care Centre (Rare Care, Clinical Centre of Expertise for Rare and Undiagnosed Diseases) is based at Perth Children's Hospital. It opened in 2022 to help meet the challenges faced by children and families living with rare and undiagnosed conditions.

The Centre offers two clinical services: a WA statewide care coordination service that works across sectors, and the Nurse Navigator program.

pch.health.wa.gov.au/Our-services/Rare-Care-Centre

National research

Genomics of Rare Disease Registry (Garvan Institute of Medical Research)

This is a national registry for people who have a rare genetic condition, or one that is suspected but not yet found. By joining, families help researchers understand rare conditions better and connect with new research opportunities. Taking part is done online and starts with a short survey about your child's health and medical history. The registry can also let families know about research studies they might be able to join, including trials of new treatments.

garvan.org.au/research/clinical-trials/rare-disease-registry

International research

MyGene2

MyGene2 is a free website for families who have a rare condition. It was built by families with rare conditions, by doctors, and by researchers.

The website helps families find the gene that is causing a rare condition.

On MyGene2, families can create a free profile and choose to share things like their child's symptoms, photos, and any genetic test results.

Sharing can help in a few ways. Families may find a genetic diagnosis, get researchers to have a new look at shared genetic data if their child does not yet have a diagnosis, and connect with doctors and researchers.

They can also search for and contact other families who have the same condition or a change in the same gene. This helps families learn from each other about what to expect.

mygene2.org

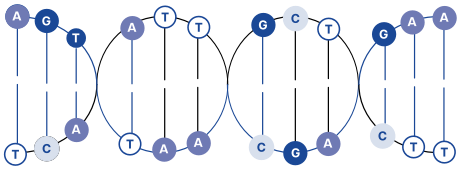


A closer look at genetics

Many, but not all, rare conditions starting in childhood have a genetic cause and so are also genetic conditions. Your local clinical genetics team can give you more personal information about genetics, but here is some general information about genetic conditions.

A genetic condition is a health condition caused by a change in a person's genes or chromosomes. We all have small differences in our genes. Some make no difference, others can cause health conditions.

Some genetic conditions are passed down from a parent. Others happen for the first time in a child.



DNA

DNA is the special code inside every cell of your child's body. DNA uses four chemical letters (A, T, G, C) in different orders, a bit like how 26 letters make all the words in a book. Every cell has the same complete set of DNA, and your child's DNA is unique to them.

Genes

Genes are the instructions for how our bodies grow and work. Each gene is like one recipe in a recipe book. Your child has more than 20,000 genes. Each one is a pattern of DNA letters. Some genes set eye colour, some help the heart work, some help the brain develop. Most genes come in pairs, with one copy from each parent.

Chromosomes

Chromosomes are like the chapters in your child's recipe book. Most children have 46 chromosomes, in 23 pairs, holding around 20,000 pairs of genes. Pairs give most genes a backup copy. If one copy does not work, the other can often do the job.

You can learn more about DNA, genes and chromosomes on the Centre for Genetics Education website: genetics.edu.au



Where to get reliable information about genetics and rare conditions

Rare Voices Australia.
The national peak body for Australians living with a rare condition. The RARE Portal gives families step-by-step guidance on what to do if a rare disease is diagnosed or suspected.

rarevoices.org.au



Centre for Genetics Education.
Australian website with clear, reliable information about genetics, genetic conditions, testing, pregnancy, and genetic services.

genetics.edu.au



National Centre for Indigenous Genomics.
A leading centre that uses genomics to improve the health and wellbeing of Indigenous Australians. It offers a growing collection of culturally appropriate resources about genetics and genomics, including videos and fact sheets.

ncig.anu.edu.au/genetic-resources



GeneEQUAL.
GeneEQUAL is an inclusive program which has a growing range of Easy Read factsheets and videos made with people with intellectual disability about genetic health care and genetic conditions.

geneequal.com/



MedlinePlus Genetics.
Easy-to-read information about genetic conditions, genes, chromosomes and health.

medlineplus.gov/genetics



NORD.
The National Organization for Rare Disorders. A US-based database with reliable information on more than 1,200 rare diseases, including symptoms, causes, diagnosis, treatment and support.

rarediseases.org



Unique.
An international charity for those affected by rare chromosome and gene differences. Provides condition guides and links to other families.

rarechromo.org



Rare Diseases NSW.
A centre of expertise for rare and undiagnosed conditions. Includes links to clinical services, current research and resources.

rarediseasesnsw.au



Being prepared

Emergency information

Even simple preparation helps in an emergency. Keep a short medical summary on hand: your child's name, date of birth, Medicare number, main medical concerns, current medications, allergies, what helps when they are distressed, and your contact number.

You may like to talk to your doctor about completing a rare disease passport. You can find more information about this here:

rarediseasesnsw.au

Planning for the future

Some gentle future planning can bring peace of mind. Around age 14, families often start thinking about the move from children's services to adult services. There is a network of transition services to support families with this move – more details below:

Transition care resources

Transition Care Network (NSW Health Agency for Clinical Innovation)

Key principles and resources to support successful transition from paediatric to adult services in New South Wales and the Australian Capital Territory.

aci.health.nsw.gov.au/networks/transition-care

Healthy WA

My Health in My Hands (WA Health): A three-minute animation aimed at young people residing in Western Australia aged 12 to 18 years to provide support as they become more independent with their health care. Includes a transition readiness checklist for young people and a transition readiness checklist for parents and carers of young people.

health.wa.gov.au/Articles/A_E/Child-and-Youth-Health-Network

Queensland

Nurse Navigator Service (Queensland Health)

Helps families of children with complex conditions navigate services across the health care system, including related to transition.

childrens.health.qld.gov.au/service-nurse-navigators/

Transition care services

New South Wales

NSW Transition Care Network Service (NSW Health Agency for Clinical Innovation)

A New South Wales service offering practical help for young people with chronic health conditions transitioning to adult services.

aci.health.nsw.gov.au/networks/transition-care/service

Examples of services (NSW Health Agency for Clinical Innovation)

List of transition clinics and services available in New South Wales to support young people, their families, and carers through transition.

aci.health.nsw.gov.au/networks/transition-care/resources/key-principles/transition-support-services

Trapeze (Sydney Children's Hospitals Network)

Transition from paediatric to adult health for young patients with chronic conditions.

schn.health.nsw.gov.au/trapeze-sydney-childrens-hospitals-networks-health-transition-service

Western Australia

Western Australia Child and Adolescent Health Service (Perth Children's Hospital, Western Australia)

Transition to adult health care provides information and resources for adolescents and their families.

pch.health.wa.gov.au/For-patients-and-visitors/Transition-to-adult-healthcare

Victoria

Transition Support Service (Royal Children's Hospital in Melbourne, Victoria)

Assists young people with chronic medical conditions and/or disabilities and their parents and carers to transition and transfer to adult care from the age of 15 onwards.

rch.org.au/transition/

The Royal Melbourne Hospital Young Adults Transition Service (Royal Melbourne Hospital, Victoria)

Provides specialist consultation to adults with congenital disabilities who require ongoing monitoring and care with regards to their physical wellbeing.

thermh.org.au/services/community-services/young-adults-transition-service

The road ahead

Your child's diagnosis will shape many parts of your lives. It does not change their worth, what they can do, or the love they bring to the people around them.

There may be times of uncertainty, grief or exhaustion, and it is important to acknowledge these feelings. There can also be deep joy, unexpected strengths and meaningful connections.

You do not need to have everything figured out at once. No family begins this journey with a clear map. Most families learn over time. They become more confident and better at noticing progress in all its forms.

Seek support when needed, and accept help when it is offered.

Trust your knowledge of your child. You are the expert in who they are, what comforts them, what excites them, and what helps them feel safe.

As research, awareness and specialist services keep growing, families have better information, stronger networks and more support than ever before. There is still much to learn, but progress is happening. Families, clinicians, researchers and communities are all part of that progress.

With the right support, advocacy and care, children with rare conditions can thrive in ways that are uniquely their own.



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3. Rare Voices Australia. What is a rare disease? Available from rarevoices.org.au/what-is-a-rare-disease/
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This document was written by Nika Kulaweera and Jessie Ballentine, in consultation with the Rare Diseases NSW and Gene2Care teams.



Nika Kulaweera
Clinical Nurse Specialist

The Sydney Children's Hospitals Network



Jessie Ballentine
Social Worker

The Sydney Children's Hospitals Network



Associate Professor Elizabeth Palmer
Clinical Geneticist

The Sydney Children's Hospitals Network & UNSW Sydney



Dr Lauren Kelada
Psychosocial Researcher

UNSW Sydney



Natalie Roberts
Program Manager

UNSW Sydney



Maddison Smith
PhD Candidate

UNSW Sydney



Rebecca Vink
Genetic Counsellor

The Sydney Children's Hospitals Network



Krista Recsei
Program Manager

UNSW Sydney



Kristine Pierce
Consumer Advocate

UNSW Sydney



Elisha Swainson
Genetic Counsellor

UNSW Sydney



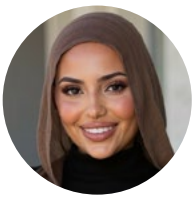
Jackson Horton
Medical Student and Research Assistant

UNSW Sydney



Samantha Flecha
Executive Officer

UNSW Sydney



Dalia Metwally
PhD Candidate

UNSW Sydney



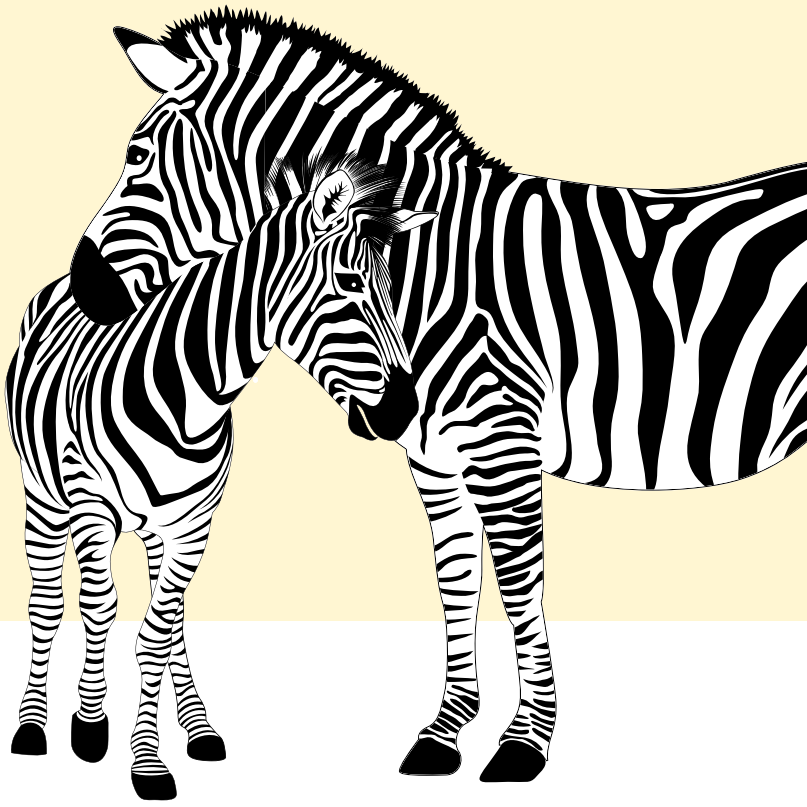
Bethany Wadling
Genetic Counsellor

Royal Hospital for Women



Emma Bonser
Chief Executive Officer

Genetic Alliance Australia



Rare Diseases
NSW